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SUBCLASS CLASS



Mouse Sox9 Sequence

AGTTTCAGTC CAGGAACTTT TCTTTGCAAG AGAGACGAGG TGCAAGTGGC CCCGGTTTCG TTCTCTGTTT TCCCTCCCTC CTCCTCCGCT CCGACTCGCC 51 101 TTCCCCGGGT TTAGAGCCGG CAGCTGAGAC CCGCCACCCA GCGCCTCTGC TAAGTGCCCG CCGCCGCAGC CCGGTGACGC GCCAACCTCC CCGGGAGCCG 151 201 TTCGCTCGGC GTCCGCGTCC GGGCAGCTGA GGGAAGAGGA GCCCCAGCCG CCGCGGCTTC TCGCCTTTCC CGGCCACCCG CCCCTGCCC CGGGCTCGCG 251 301 TATGAATCTC CTGGACCCCT TCATGAAGAT GACCGACGAG CAGGAGAAGG GCCTGTCTGG CGCCCCCAGC CCCACCATGT CGGAGGACTC GGCTGGTTCG 351 401 CCCTGTCCCT CGGGCTCCGG CTCGGACACG GAGAACACCC GGCCCCAGGA 451 GAACACCTTC CCCAAGGGCG AGCCGGATCT GAAGAAGGAG AGCGAGGAAG ATAAGTTCCC CGTGTGCATC CGCGAGGCGG TCAGCCAGGT GCTGAAGGGC 501 TACGACTGGA CGCTGGTGCC CATGCCCGTG CGCGTCAACG GCTCCAGCAA 551 GAACAAGCCA CACGTCAAGC GACCCATGAA CGCCTTCATG GTGTGGGCGC 601 AGGCTGCGCG CAGGAAGCTG GCAGACCAGT ACCCGCATCT GCACAACGCG 651 701 GAGCTCAGCA AGACTCTGGG CAAGCTCTGG AGGCTGCTGA ACGAGAGCGA 751 GAAGAGCCC TTCGTGGAGG AGGCGGAGCG GCTGCGCGTG CAGCACAAGA 801 AAGACCACCC CGATTACAAG TACCAGCCCC GGCGGAGGAA GTCGGTGAAG 851 AACGGACAAG CGGAGGCCGA AGAGGCCACG GAACAGACTC ACATCTCTCC 901 TAATGCTATC TTCAAGGCGC TGCAAGCCGA CTCCCCACAT TCCTCCTCCG GCATGAGTGA GGTGCACTCC CCGGGCGAGC ACTCTGGGCA ATCTCAGGGT 951 1001 CCGCCGACCC CACCCACCAC TCCCAAAACC GACGTGCAAG CTGGCAAAGT 1051 TGATCTGAAG CGAGAGGGGC GCCCTCTGGC AGAGGGGGGC AGACAGCCCC 1101 CCATCGACTT CCGCGACGTG GACATCGGTG AACTGAGCAG CGACGTCATC TCCAACATTG AGACCTTCGA CGTCAATGAG TTTGACCAAT ACTTGCCACC 1151 1201 CAACGGCCAC CCAGGGGTTC CGGCCACCCA CGGCCAGGTC ACCTACACTG GCAGTTACGG CATCAGCAGC ACCGCACCCA CCCCTGCGAC CGCGGGCCAC

Figure 1(a)

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1301	GTGTGGATGT	CGAAGCAGCA	GGCGCCGCCC	CCTCCTCCGC	AGCAGCCTCC
1351	GCAGGCCCCG	CAAGCCCCAC	AGGCGCCTCC	GCAGCAGCAA	GCACCCCCGC
1401	AGCAGCCGCA	GGCACCCCAG	CAGCAGCAGG	CACACACGCT	CACCACGCTG
1451	AGCAGCGAGC	CAGGCCAGTC	CCAGCGAACG	CACATCAAGA	CGGAGCAGCT
1501	GAGCCCCAGC	CACTACAGGG	AGCAGCAGCA	GCACTCCCCG	CAACAGATCT
1551	CCTACAGCCC	CTTCAACCTT	CCTCACTACA	GGCCCTCCTA	CCCGCCCATC
1601	ACCCGTTCGG	AATACGACTA	CGCTGACCAT	CAGAACTCCG	GCTCCTACTA
1651	CAGTCACGCA	GCCGGCCAGG	GCTCAGGGCT	CTACTCCACC	TTCACTTACA
1701	TGAACCCCGC	GCAGCGCCCC	ATGTACACCC	CCATCGGTGA	CACCTCCGGG
1751	GTCCCTTCCA	TCCCGCAGAC	CCACAGCCCG	CAGGACTGGG	AACAACCAGT
1801	CTACACACAG	GTCACCAGAC	CCTGAGAAGA	GAAAAGCTAT	GGTGACAGAG
1851	CTGATCTTTT	${\tt TTTTTTTTTT}$	TTTTTAAAGA	AGAAAAGAAA	GAAACGAAAA
1901	AGAAAAAGCT	GAAGGAAATC	AAGAACCAAT	TGAAATTCCT	TTGGACACTT
1951	TTTTTTTTTTTTT	CCTTTCGTTA	ATTTTTAAAA	GACATGTAAA	GGAAGGTAAC
2001	GATTGCTGGG	CATTCCAGGA	GAGAGACTTT	AAGACTTTGT	CTGAGCTCAT
2051	GACAACATAT	TGCAAATGGC	CGGGCCACTC	GTGGCCAGAC	GGACAGCACT
2101	CCTGGCCAGA	TGGACCCACC	AGTATCAGCG	AGGAGGGGCT	TGTCTCCTTC
2151	AGAGTTAACA	TGGAGGACGA	TTGGAGAATC	TCCCTGCCTG	TTTGGACTTT
2201	GTAATTATTT	TTTAGCCGTA	ATTAAAGAAA	AAAAAAGTCC	ААААААА

Figure 1(b)

1

APPROVED O.G. FIG.
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Mouse Sox-9 amino acid sequence

Met Asn Leu Leu Asp Pro Phe Met Lys Met Thr Asp Glu Gln Glu Lys Gly Leu Ser Gly Ala Pro Ser Pro Thr Met Ser Glu Asp Ser Ala Gly Ser Pro Cys Pro Ser Gly Ser Gly Ser Asp Thr Glu Asn Thr Arg Pro Gln Glu Asn Thr Phe Pro Lys Gly Glu Pro Asp Leu Lys Lys Glu Ser Glu Glu Asp Lys Phe Pro Val Cys Ile Arg Glu Ala Val Ser Gln Val Leu Lys Gly Tyr Asp Trp Thr Leu Val Pro Met Pro Val Arg Val Asn Gly Ser Ser Lys Asn Lys Pro His Val Lys Arg Pro Met Asn Ala Phe Met Val Trp Ala Gln Ala Ala Arg Arg Lys Leu Ala Asp Gln Tyr Pro His Leu His Asn Ala Glu Leu Ser Lys Thr Leu Gly Lys Leu Trp Arg Leu Leu Asn Glu Ser Glu Lys Arg Pro Phe Val Glu Glu Ala Glu Arg Leu Arg Val Gln His Lys Lys Asp His Pro Asp Tyr Lys Tyr Gln Pro Arg Arg Arg Lys Ser Val Lys Asn Gly Gln Ala Glu Ala Glu Glu Ala 185 Thr Glu Gln Thr His Ile Ser Pro Asn Ala Ile Phe Lys Ala Leu Gln Ala Asp Ser Pro His Ser Ser Ser Gly Met Ser Glu Val His Ser Pro Gly Glu His Ser Gly Gln Ser Gln Gly Pro Pro Thr Pro Pro Thr Thr 235 Pro Lys Thr Asp Val Gln Ala Gly Lys Val Asp Leu Lys Arg Glu Gly 250 Arg Pro Leu Ala Glu Gly Gly Arg Gln Pro Pro Ile Asp Phe Arg Asp

Figure 1(c)

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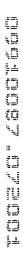
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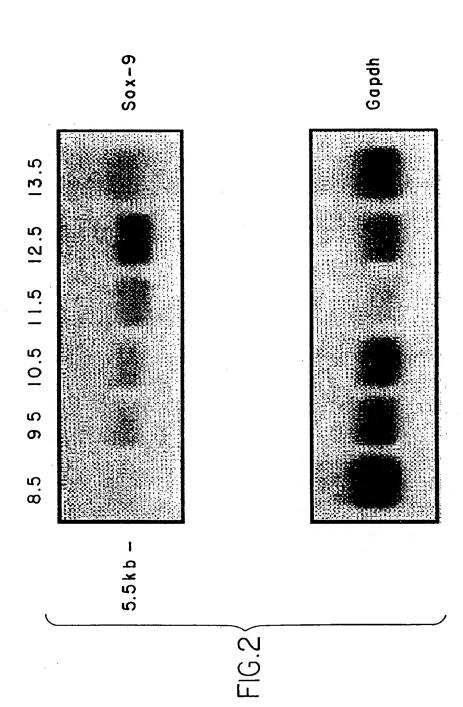
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APPROVED O.G. FIG.
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Val Asp Ile Gly Glu Leu Ser Ser Asp Val Ile Ser Asn Ile Glu Thr Phe Asp Val Asn Glu Phe Asp Gln Tyr Leu Pro Pro Asn Gly His Pro Gly Val Pro Ala Thr His Gly Gln Val Thr Tyr Thr Gly Ser Tyr Gly 310 Ile Ser Ser Thr Ala Pro Thr Pro Ala Thr Ala Gly His Val Trp Met Ser Lys Gln Gln Ala Pro Pro Pro Pro Gln Gln Pro Pro Gln Ala Pro Gln Ala Pro Gln Ala Pro Pro Gln Gln Gln Ala Pro Pro Gln Gln 360 Pro Gln Ala Pro Gln Gln Gln Ala His Thr Leu Thr Thr Leu Ser Ser Glu Pro Gly Gln Ser Gln Arg Thr His Ile Lys Thr Glu Gln Leu 390 Ser Pro Ser His Tyr Arg Glu Gln Gln Gln His Ser Pro Gln Gln Ile Ser Tyr Ser Pro Phe Asn Leu Pro His Tyr Arg Pro Ser Tyr Pro Pro 425 Ile Thr Arg Ser Glu Tyr Asp Tyr Ala Asp His Gln Asn Ser Gly Ser Tyr Tyr Ser His Ala Ala Gly Gln Gly Ser Gly Leu Tyr Ser Thr Phe 450 Thr Tyr Met Asn Pro Ala Gln Arg Pro Met Tyr Thr Pro Ile Gly Asp 470 Thr Ser Gly Val Pro Ser Ile Pro Gln Thr His Ser Pro Gln Asp Trp 485 490 Glu Gln Pro Val Tyr Thr Gln Val Thr Arg Pro

Figure 1(d)





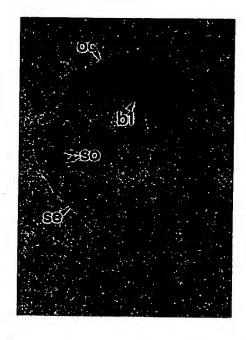


FIG.3a



FIG.3b

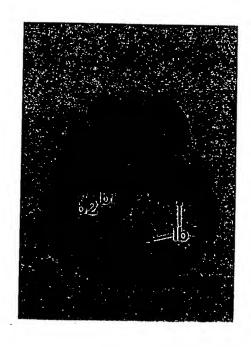


FIG.3c

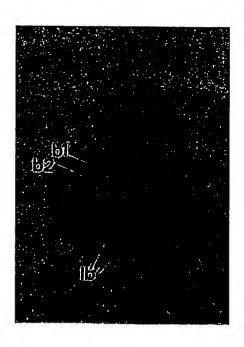


FIG.3d

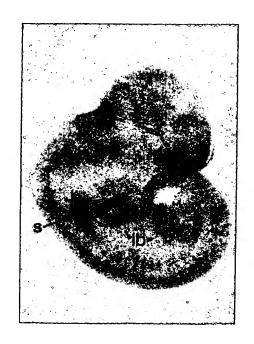


FIG.3e

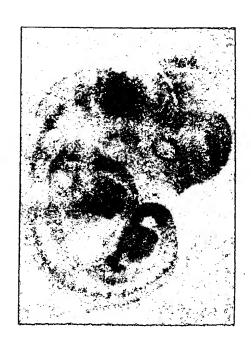


FIG.3f



FIG.3g

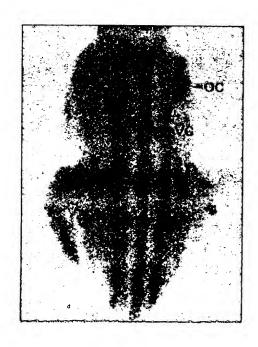


FIG.3h



FIG.3i

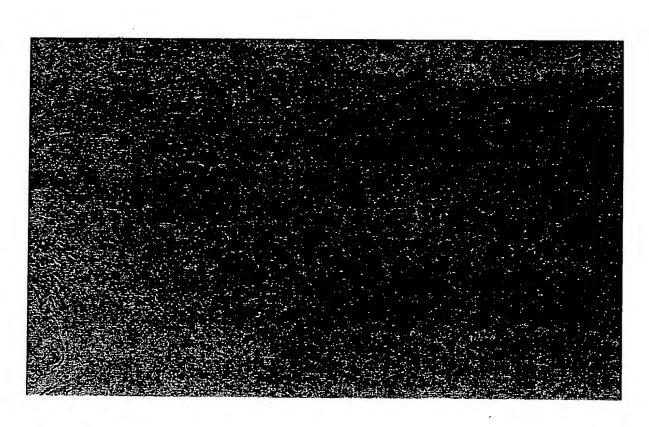


FIG.4

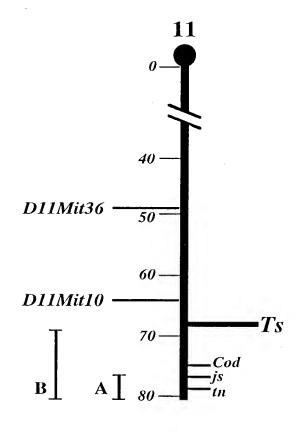
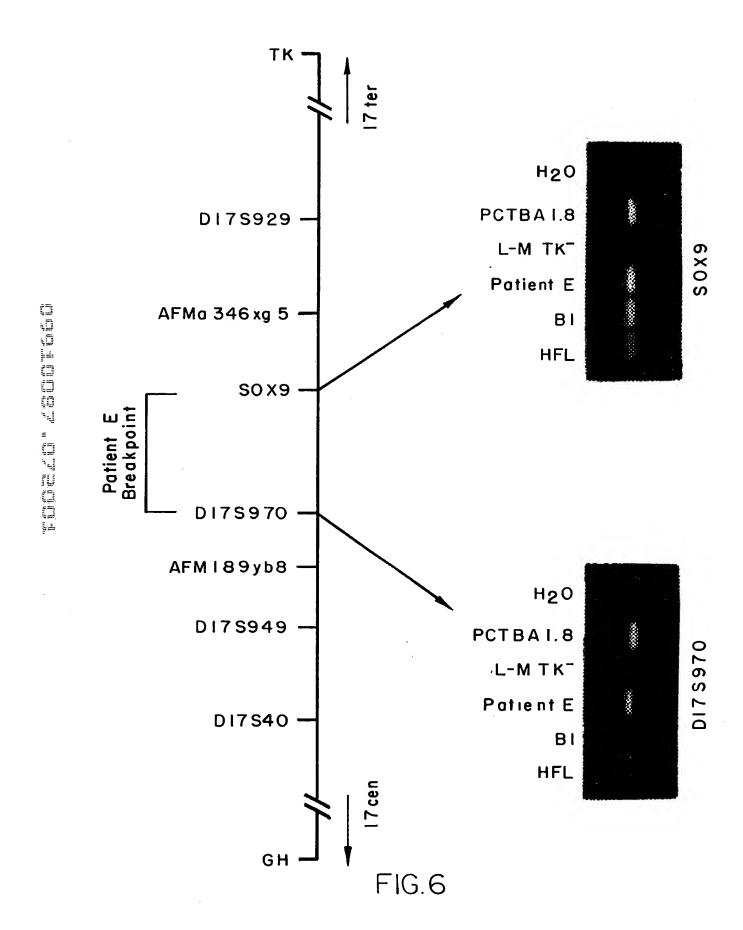
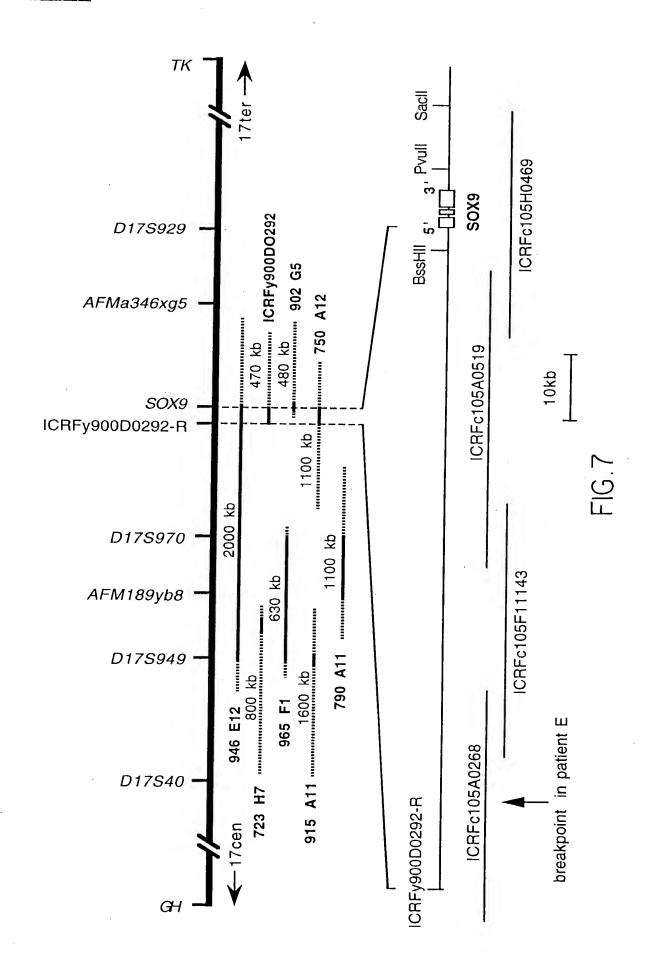


FIG.5

APPROVED	O.G. FIG.						
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APPROVED	O.G. FIG.						
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CGGAGCTCGA AACTGACTGG AAACTTCAGT GGCGCGGAGA CTCGCCAGTT TCAACCCCGG AAACTTTTCT TTGCAGGAGG AGAAGAGAAG GGGTGCAAGC GCCCCACTT TTGCTCTTTT
TCCTCCCCTC CTCCTCCTCT CCAATTCGCC TCCCCCCACT TGGAGCGGGC AGCTGTGAAC
TGGCCACCCC GCGCTTCCT AAGTGCTCGC CGCGGTAGCC GGCCGACGCG CCAGCTTCCC
CGGGAGCCGC TTGCTCCGCA TCCGGGCAGC CGAGGGGAGA GGAGCCCGCG CCTCGAGTCC
CCGAGCCGCC GCGCTTCTC GCCTTTCCCG GCCACCAGCC CCCTGCCCCG GGCCCGCGTA
TGAATCTCCT GGACCCCTTC ATGAAGATGA CCGACGAGCA GGAGAAGGGC CTGTCCGGCG CCCCCAGCCC CACCATGTCC GAGGACTCCG CGGGCTCGCC CTGCCCGTCG GGCTCCGGCT CGGACACCGA GAACACGCGG CCCCAGGAGA ACACGTTCCC CAAGGGCGAG CCCGATCTGA AGAAGGAGAG CGAGGAGGAC AAGTTCCCCG TGTGCATCCG CGAGGCGGTC AGCCAGGTGC TCAAAGGCTA CGACTGGACG CTGGTGCCCA TGCCGGTGCG CGTCAACGGC TCCAGCAAGA ACAAGCCGCA CGTCAAGCGG CCCATGAACG CCTTCATGGT GTGGGCGCAG GCGGCGCGCA GGAAGCTCGC GGACCAGTAC CCGCACTTGC ACAACGCCGA GCTCAGCAAG ACGCTGGGCA AGCTCTGGAG ACTTCTGAAC GAGAGCGAGA AGCGGCCCTT CGTGGAGGAG GCGGAGCGGC TGCGCGTGCA GCACAAGAAG GACCACCCGG ATTACAAGTA CCAGCCGCGG CGGAGGAAGT CGGTGAAGAA CGGGCAGGCG GAGGCAGAGG AGGCCACGGA GCAGACGCAC ATCTCCCCCA ACGCCATCTT CAAGGCGCTG CAGGCCGACT CGCCACACTC CTCCTCCGGC ATGAGCGAGG TGCACTCCCC CGGCGAGCAC TCGGGGCCAAT CCCAGGGCCC ACCGACCCCA CCCACCACCC CCAAAACCGA CGTGCAGCCG GGCAAGGCTG ACCTGAAGCG AGAGGGGCGC CCCTTGCCAG AGGGGGGCAG ACAGCCCCCT ATCGACTTCC GCGACGTGGA CATCGGCGAG CTGAGCAGCG ACGTCATCTC CAACATCGAG ACCTTCGATG TCAACGAGTT TGACCAGTAC CTGCCGCCCA ACGGCCACCC GGGGGTGCCG GCCACGCACG GCCAGGTCAC CTACACGGGC AGCTACGGCA TCAGCAGCAC CGCGGCCACC CCGGCGAGCG CGGGCCACGT GTGGATGTCC AAGCAGCAGG CGCCGCCGC ACCCCGCAG CAGCCCCCAC AGGCCCCGCC GGCCCCGCAG GCGCCCCCGC AGCCGCAGGC GGCGCCCCCA CAGCAGCCGG CGGCACCCCC GCAGCAGCCA CAGGCGCACA CGCTGACCAC GCTGAGCAGC GAGCAGCCG CGGCACCCC GCAGCAGCA CAGGCGCACAC
CGCTGACCAC GCTGAGCAGC GAGCCGGGC AGCCCCACACAC AAGACGGAGC
AGCTGAGCCC CAGCCACTAC AGCGAGCAGC AGCAGCACTC GCCCCAACAG ATCGCCTACA
GCCCCTTCAA CCTCCCACAC TACAGCCCCT CCTACCCGCC CATCACCCGC TCACAGTACG ACTACACCGA CCACCAGAAC TCCAGCTCCT ACTACAGCCA CGCGGCAGGC CAGGGCACCG GCCTCTACTC CACCTTCACC TACATGAACC CCGCTCAGCG CCCCATGTAC ACCCCCATCG CCGACACCTC TGGGGTCCCT TCCATCCCGC AGACCCACAG CCCCCAGCAC TGGGAACAAC CCGTCTACAC ACAGCTCACT CGACCTTGAG GAGGCCTCCC ACGAAGGGCG ACGATGGCCG TGTACTGTGT ATGATTCATT ACCATTTTGA GGGGATTTAT ACATATTTTT AGATAAAATT AAATGCTCTT ATTTTTCCAA CAGCTAAACT ACTCTTAGTT GAACAGTGTG CCCTAGCTTT TCTTGCAACC AGAGTATTTT TGTACAGATT TGCTTTCTCT TACAAAAAGA AAAAAAAAT CCTGTTGTAT TAACATTTAA AAACAGAATT GTGTTATGTG ATCAGTTTTG GGGGTTAACT TTGCTTAATT CCTCAGGCTT TGCGATTTAA GGAGGAGCTG CCTTAAAAAA AAATAAAGGC CTTATTTTGC AATTATGGGA GTAAACAATA GTCTAGAGAA GCATTTGGTA AGCTTTATGA TATATATT TTTTAAAGAA GAGAAAAACA CCTTGAGCCT TAAAACGGTG CTGCTGGGAA ACATTTGCAC TCTTTTAGTG CATTTCCTCC TGCCTTTGCT TGTTCACTGC AGTCTTAAGA AAGAGGTAAA AGGCAAGCAA AGGAGATGAA ATCTGTTCTG GGAATGTTTC AGCAGCCAAT AAGTGCCCGA GCACACTGCC CCCGGTTGCC TGCCTGGGCC CCATGTGGAA GGCAGATGCC TGCTCGCTCT GTCACCTGTG CCTCTCAGAA CACCAGCAGT TAACCTTCAA GACATTCCAC

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TTGCTAAAAT TATTTATTTT GTAAGGAGAG GTTTTAATTA AAACAAAAA AAATTCTTTT TTTTTTTTT TTTTCCAATT TTACCTTCTT TAAAATAGGT TGTTGGAGCT TTCCTCAAAG GGTATGGTCA TCTGTTGTTA. AATTATGTTC TTAACTGTAA CCAGTTTTTT TTTATTTATC TCTTTAATCT TTTTTATTAT TAAAAGCAAG TTTCTTTGTA TTCCTCACCC TAGATTTGTA TAAATGCCTT TTTGTCCATC CCTTTTTTCT TTGTTGTTTT TGTTGAAAAC AAACTGGAAA CTTGTTTCTT TTTTTGTATA AATGAGAGAT TGCAAATGTA GTGTATCACT GAGTCATTTG CAGTGTTTTC TGCCACAGAC CTTTGGGCTG CCTTATATTG TGTGTGTGTG TGGGTGTGTG TGTGTTTTGA CACAAAACA ATGCAAGCAT GTGTCATCCA TATTTCTCTA CATCTTCTCT TGGAGTGAGG GAGGCTACCT GGAGGGGATC AGCCCACTGA CAGACCTTAA TCTTAATTAC TGCTGTGGCT AGAGAGTTTG AGGATTGCTT TTTAAAAAAG ACAGCAAACT TTTTTTTTA TTTAAAAAA GATATATAA CAGTTTTAGA AGTCAGTAGA ATAAAATCTT AAAGCACTCA TAATATGGCA TCCTTCAATT TCTGTATAAA AGCAGATCTT TTTAAAAAAG ATACTTCTGT AACTTAAGAA ACCTGGCATT TAAATCATAT TTTGTCTTTA GGTAAAAGCT TTGGTTTGTG TTCGTGTTTT GTTTGTTTCA CTTGTTTCCC TCCCAGCCCC AAACCTTTTG TTCTCTCCGT ATATACATTG CATTAAAAAG AAA

Figure 8a(2)

APPROVED	O.G. FIG.						
BY	CLASS	SUBCLASS					
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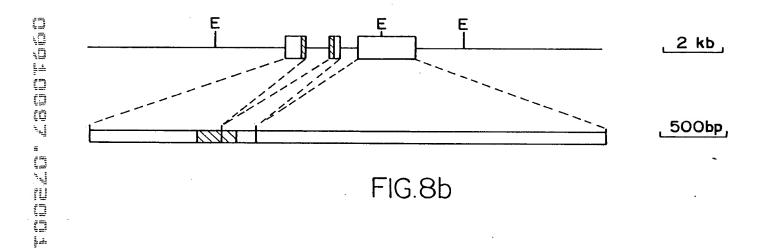
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Gly	Leu	Ser	Gly 20	Ala	Pro	Ser	Pro	Thr 25		Ser	Glu	Asp	Ser 30		Gly
Ser	Pro	Cys 35	Pro	Ser	Gly	Ser	Gly 40	Ser	Asp	Thr	Glu	Asn 45		Arg	Pro
	50				Pro	55					60		_		
65					Pro 70					75					80
				85	Trp				90				_	95	-
			100		Lys			105					110		
		115			Ala		120					125			
	130				Glu	135					140				
145					Glu 150					155					160
				165	Lys				170					175	
			180		Val			185					190		
		195			Ile		200					205			
	210				Ser	215					220				Pro
225					Gln 230					235					Thr 240
Pro				245	Gln				250					255	
Arg			260		Gly	•		265					270		
Val		275			Leu		280			•		285			
	290				Phe	295					300				
Gly 305					His 310					315				_	320
Ile				325	Ala				330			•		335	
Ser		_	340		Pro			345					350		
		355			Ala		360					365			
Gln	Pro 370	Ala	Ala	Pro	Pro	Gln 375	Gln	Pro	Gln	Ala	His 380	Thr	Leu	Thr	Thr

Figure 8a(3)

APPROVEB O.G. FIG.
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Leu Ser Ser Glu Pro Gly Gln Ser Gln Arg Thr His Ile Lys Thr Glu Gln Leu Ser Pro Ser His Tyr Ser Glu Gln Gln His Ser Pro Gln Gln Ile Ala Tyr Ser Pro Phe Asn Leu Pro His Tyr Ser Pro Ser Tyr Pro Pro Ile Thr Arg Ser Gln Tyr Asp Tyr Thr Asp His Gln Asn Ser Ser Ser Tyr Tyr Ser His Ala Ala Gly Gln Gly Thr Gly Leu Tyr Ser Thr Phe Thr Tyr Met Asn Pro Ala Gln Arg Pro Met Tyr Thr Pro Ile Ala Asp Thr Ser Gly Val Pro Ser Ile Pro Gln Thr His Ser Pro Gln His Trp Glu Gln Pro Val Tyr Thr Gln Leu Thr Arg Pro

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BY	CLASS	SUBCLASS		
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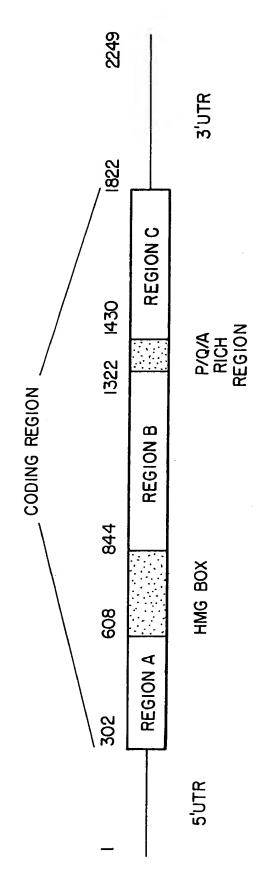
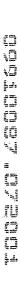


FIG.9



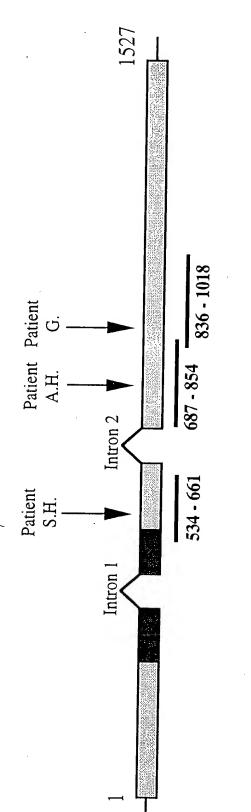
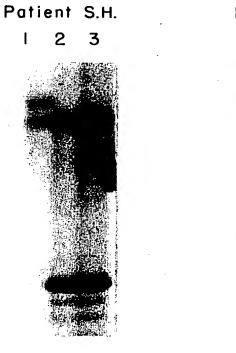
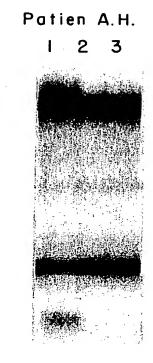


FIG. 100





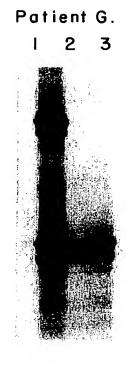


FIG.IOb

Patient A.H.

Mutated Normal allele allele

Mutated

Normal

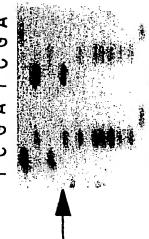
Patient S.H.

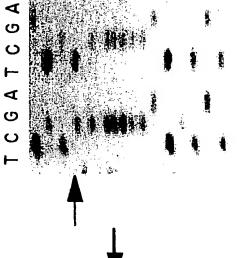
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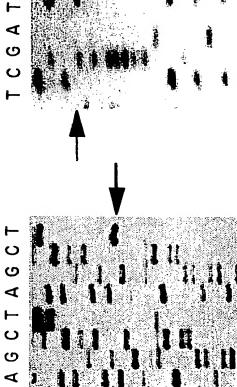
allele

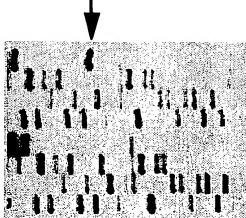
Patient G. allele

TCGATCGA Normal Mutated allele









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